



acromicric dysplasia

Acromicric dysplasia is a condition characterized by severely short stature, short limbs, stiff joints, and distinctive facial features.

Newborns with acromicric dysplasia are of normal size, but slow growth over time results in short stature. The average height of adults with this disorder is about 4 feet, 2 inches for women and 4 feet, 5 inches for men. The long bones of the arms and legs, and the bones in the hands and feet, are shorter than would be expected for the individual's height. Other skeletal features that occur in this disorder include slowed mineralization of bone (delayed bone age), abnormally shaped bones of the spine (vertebrae), and constrained movement of joints. Affected individuals often develop carpal tunnel syndrome, which is characterized by numbness, tingling, and weakness in the hands and fingers. A misalignment of the hip joints (hip dysplasia) can also occur in this disorder. These skeletal and joint problems may require treatment, but most affected individuals have few limitations in their activities.

Children with acromicric dysplasia may have a round face, sharply defined eyebrows, long eyelashes, a bulbous nose with upturned nostrils, a long space between the nose and upper lip (philtrum), and a small mouth with thick lips. These facial differences become less apparent in adulthood. Intelligence is unaffected in this disorder, and life expectancy is generally normal.

Frequency

Acromicric dysplasia is a rare disorder; its prevalence is unknown.

Genetic Changes

Acromicric dysplasia is caused by mutations in the *FBN1* gene, which provides instructions for making a large protein called fibrillin-1. This protein is transported out of cells into the extracellular matrix, which is an intricate lattice of proteins and other molecules that forms in the spaces between cells. In this matrix, molecules of fibrillin-1 attach (bind) to each other and to other proteins to form threadlike filaments called microfibrils. The microfibrils become part of the fibers that provide strength and flexibility to connective tissues, which support the bones, skin, and other tissues and organs. Additionally, microfibrils store molecules called growth factors, including transforming growth factor beta (TGF- β), and release them at various times to control the growth and repair of tissues and organs throughout the body.

Most of the *FBN1* gene mutations that cause acromicric dysplasia change single protein building blocks in the fibrillin-1 protein. The mutations result in a reduction and

disorganization of the microfibrils. Without enough normal microfibrils to store TGF- β , the growth factor is abnormally active. These effects likely contribute to the physical abnormalities that occur in acromicric dysplasia, but the mechanisms are unclear.

Inheritance Pattern

Acromicric dysplasia is an autosomal dominant condition, which means one copy of the altered gene in each cell is sufficient to cause the disorder. Most cases result from new mutations in the gene and occur in people with no history of the disorder in their family. In other cases, an affected person inherits the mutation from one affected parent.

Other Names for This Condition

- ACMICD

Diagnosis & Management

These resources address the diagnosis or management of acromicric dysplasia:

- Genetic Testing Registry: Acromicric dysplasia
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0265287/>

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>

Additional Information & Resources

MedlinePlus

- Health Topic: Bone Diseases
<https://medlineplus.gov/bonediseases.html>

Genetic and Rare Diseases Information Center

- Acromicric dysplasia
<https://rarediseases.info.nih.gov/diseases/7/acromicric-dysplasia>

Educational Resources

- Disease InfoSearch: Acromicric Dysplasia
<http://www.diseaseinfosearch.org/Acromicric+Dysplasia/167>
- MalaCards: acromicric dysplasia
http://www.malacards.org/card/acromicric_dysplasia
- Merck Manual Consumer Version: Osteochondrodysplasias
<http://www.merckmanuals.com/home/children-s-health-issues/hereditary-connective-tissue-disorders/osteochondrodysplasias>
- Orphanet: Acromicric dysplasia
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=969

Patient Support and Advocacy Resources

- Human Growth Foundation
<http://hgfound.org/>
- Little People of America
<http://www.lpaonline.org/>
- National Organization for Rare Disorders (NORD)
<http://rarediseases.org/rare-diseases/acromicric-dysplasia/>
- The MAGIC Foundation
<https://www.magicfoundation.org/>

Genetic Testing Registry

- Acromicric dysplasia
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0265287/>

Scientific articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28acromicric+dysplasia%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

OMIM

- ACROMICRIC DYSPLASIA
<http://omim.org/entry/102370>

Sources for This Summary

- Faivre L, Le Merrer M, Baumann C, Polak M, Chatelain P, Sulmont V, Cousin J, Bost M, Cordier MP, Zackai E, Russell K, Finidori G, Pouliquen JC, Munnich A, Maroteaux P, Cormier-Daire V. Acromicric dysplasia: long term outcome and evidence of autosomal dominant inheritance. *J Med Genet.* 2001 Nov;38(11):745-9.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/11694546>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1734753/>
 - Klein C, Le Goff C, Topouchian V, Odent S, Violas P, Glorion C, Cormier-Daire V. Orthopedics management of acromicric dysplasia: follow up of nine patients. *Am J Med Genet A.* 2014 Feb; 164A(2):331-7. doi: 10.1002/ajmg.a.36139. Epub 2013 Dec 11.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/24339047>
 - Le Goff C, Mahaut C, Wang LW, Allali S, Abhyankar A, Jensen S, Zylberberg L, Collod-Beroud G, Bonnet D, Alanay Y, Brady AF, Cordier MP, Devriendt K, Genevieve D, Kiper PÖ, Kito H, Krakow D, Lynch SA, Le Merrer M, Mégarbane A, Mortier G, Odent S, Polak M, Rohrbach M, Sillence D, Stolte-Dijkstra I, Superti-Furga A, Rimoin DL, Topouchian V, Unger S, Zabel B, Bole-Feysot C, Nitschke P, Handford P, Casanova JL, Boileau C, Apte SS, Munnich A, Cormier-Daire V. Mutations in the TGF β binding-protein-like domain 5 of FBN1 are responsible for acromicric and geleophysic dysplasias. *Am J Hum Genet.* 2011 Jul 15;89(1):7-14. doi: 10.1016/j.ajhg.2011.05.012. Epub 2011 Jun 16.
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